

California State University, Fresno
Department of Biology presents

Improving structural variation prediction with sparsity and relatedness models



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Every time a DNA sequence duplicates, there is an opportunity for variation, such as insertions and deletions of short segments of DNA. The introduction and accumulation of genomic variation are important evolutionary drivers of phenomena such as the creation of new species as well as genetic diseases like cancer. To predict genomic variation between members of the same species, we develop a constrained-optimization method to detect germline and novel structural variants (SVs) in family lineages. We attempt to mitigate the deleterious effects of low-coverage sequences by following a maximum likelihood approach to SV prediction. Specifically, we model the noise using Poisson statistics. Unlike annotated or highly curated data, sequenced genomes of related individuals tend to suffer from errors in both sequencing and mapping. Whereas previous methods either post-process hereditary information or rely on high coverage sequencing, we incorporate data from multiple related individuals to reduce the false positive rate of prediction. Since zygosity - the number of variant copies present in an individual - affect disease susceptibility and resistance, our methods also incorporate this into variant prediction. In the context of simulated genomes, parent-child trios, and larger family lineages, our relatedness and sparsity-promoting model results in improved detection of genomic variation responsible for both diversity and disease.

For further information: www.csufresno.edu/biology or phone 278-2001

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